

Katarzyna Zabrocka
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Doug Brutlag
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Nature & Nurture: The Role of Genetics and Environment in Human Disease and Characteristics

The nature vs. nurture debate is essentially dead. Most scientists agree that human development is a result of the interaction between genetics and one's environment. However, the nature vs. nurture argument continues on because of the power struggle between its two extremes. Are we mostly human because of our genes, or because of our environment? How much does each argument contribute towards the development of a fully functioning, conscious, and rational human being? In the past sixty years, our knowledge of DNA and the human genome has grown exponentially. We have sequenced the human genome and the genomes of countless other species. We have been able to start creating therapies that are personalized according to the patient's DNA. Extensive knowledge of SNP's and how our DNA replicates have given us the opportunity to sequence human genomes faster than ever before. New fields such as pharmacogenomics are opening up to accommodate and use all this new information. But are we starting to place too much emphasis on our DNA, to the point where we ignore the important influences of our environment? DNA has in some ways become a scapegoat, allowing us to lather ourselves in unhealthy luxuries.

Genetic Basis of Disease

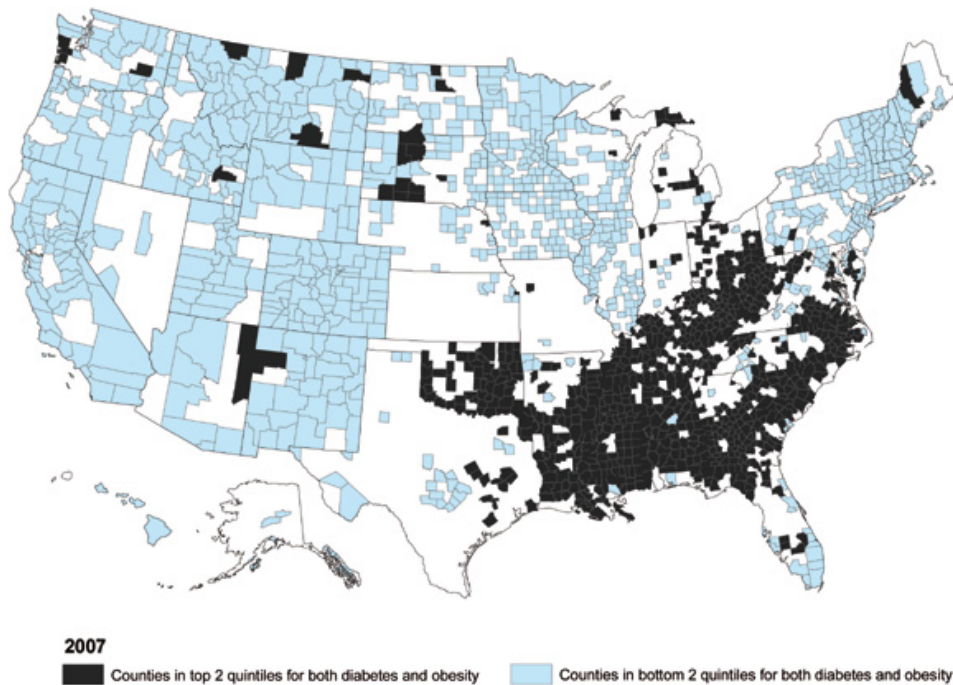
Not meaning to degrade the importance of today's scientific discoveries, genomics has allowed us to study inherited diseases more thoroughly. Websites such as Genes and Disease at NCBI [NCBI] now allow us to search genetic diseases by category (e.g. metabolic diseases) or by chromosome (e.g. 12). One such disease is Phenylketonuria, an autosomal recessive disorder caused by a mutation in the PAH gene located on 12q24.1 [Phenylketonuria]. The mutation results in a deficiency of phenylalanine hydroxylase, and thus a buildup of phenylalanine in the body, specifically in the brain. Infants with this genetic disease develop mental retardation and organ damage as they develop. The genetic basis of phenylketonuria was discovered in 1967. Though no genetic cure exists, knowledge of the basis of this disease has proven the correlation between phenylalanine and the symptoms of this disorder. With a restricted, low-phenylalanine diet, patients with phenylketonuria can live a normal and healthy life. Here we see how nurture works with nature. Nature (DNA) created a mutation and thus a genetic disorder. However, one's environment can determine the onset of the disease and the severity of its symptoms.

Many adult (or child) onset diseases, such as obesity or diabetes, have been attributed to a poor, unhealthy environment. Through the study of genomics, we have found that such diseases can also have a genetic basis. The most popular genetic theory for obesity is a "thrifty genotype", the notion that "the same genes that helped our ancestors survive occasional famines are now being challenged by environments in which food is plentiful year round." [Obesity] Genome wide association studies have also shown that early-onset and morbid adult obesity are

associated with SNP's in high-risk loci in NPC1, near MAF, and near PTER genes in European populations. [Meyre] Certain genetic diseases result in phenotype symptoms that include obesity. This includes diseases such as Prader-Willi, Bardet-Biedl, or Carpenter syndromes. [Kousta] However, to attribute all obesity cases to one's DNA is a dangerous assumption. More often than not, a genetic mutation associated with obesity is simply an indication for a predisposition for obesity. The same applies to many other chronic medical conditions such as hypertension, diabetes, and diarrheal diseases. Our environment can determine whether we develop these diseases. In western societies, where food is plentiful and high in calories, there is a high incidence of obesity. According to the CDC, more than 39.8 million American adults are overweight. The highest prevalence of obesity is found in states that value fried and high calorie foods. [Obesity] Poor diet, one high in fat and calories, is one of the biggest contributors towards obesity. Living in poor neighborhoods, where cheap food is often the unhealthiest, can also increase the risk for obesity. Diets high in sodium can cause hypertension, and eating foods high in sugar can certainly put someone at risk for type 2 diabetes. Diseases of poverty, such as diarrheal disease, can be onset by malnutrition and polluted environments, though some people may be more resistant to them than others based on their DNA. But to completely attribute such diseases to genetics is faulty – our environments and culture nurture our bodies and our DNA. If we don't acknowledge the role of our surroundings and lifestyles in the development of chronic medical conditions, we will fall into a vicious wormhole, indulging ourselves with the things we want rather than the things that are healthy for our bodies. We cannot blame our genes for poor

lifestyle choices, especially since few successful gene therapies exist. Though there are cases where genetically based medicine would be the best choice (e.g. specific test proves that genetic mutation causes the disease), a change in our environment or lifestyle is often the best medicine.

Figure 1:



[Obesity]

Unfair Lifestyle Assumptions and Judgments

Pink ribbons for breast cancer and red ribbons for AIDS awareness are plentiful in this country. What we often don't hear about is lung cancer awareness. With every "TRUTH" commercial and health class, we learn that lung cancer is a result of the decision to smoke. We are told that choosing to put cigarettes in our mouths is what causes lung cancer. Victims of lung cancer have themselves to blame.

With the influence of genomics, however, we have learned that the “victim’s” lifestyle choices are not always to blame. Some small cell lung carcinoma can be attributed to a mutation on chromosome 3 [Small Cell]. A mutation doesn’t guarantee cancer, and we still have much to learn about the role of genes in small cell lung carcinoma and other cancers. Though just as we must take heed of putting too much emphasis on genetics, if we blame the environment entirely, we might miss possible diagnoses that could lead to a cure. Furthermore, it would be wrong to always assume someone has chosen to make decisions that led him/her to their disease. Sometimes we are just dealt a bad deck of cards. With further study of the genetic and molecular basis of cancers, we can find treatments that are personalized to the cause of each patient’s symptoms.

If assuming either extreme in the nature vs. nurture debate is faulty, does that mean we should just take the middle road and say everything is caused by both nature and nurture? Matt Ridley says no. “Genes are designed to take their cues from nurture. You will have to enter a world where your genes are not puppet masters pulling the strings of your behavior but puppets at the mercy of your behavior; a world where instinct is not the opposite of learning, where environmental influences are sometimes less reversible than genetic ones, and where nature is designed for nurture.” [Ridley 4]

Human Characteristics

With about 30,000 genes in the human genome, where do we draw the line in nature vs. nurture? Our physical characteristics, excluding acquired ones, can be

attributed to genetics. We know that changes to our body, and thus our somatic cells, would not be passed on to future generations. Our height, skin color, and body build are a result of the genetic combination of our parent's DNA. However, the environment once again comes into play. Malnutrition can stunt growth, liver disease causes jaundice, and exercise routines can determine the ultimate body build. Although both our genes and environment can influence physical traits, does the same apply for human behavior?

The simple answer is yes. John Locke believed that humans are blank slates at birth, shaped by their experiences and lifestyles. Due to advances in neuroscience and genetics, we have found that not to be the full story. Our thoughts, just like the function of our bodies, have a chemical and neurological basis. Ergo, behaviors must also be linked to our genomes. One example is the link of individual empathy and stress reactivity with certain genetic variations. Individuals who had a polymorphism (rs53576) with a GG allele in the oxytocin receptor gene had a lower disposition for empathy and lower physiological and dispositional stress reactivity. [Rodrigues]. Individuals with a GA/AA allele were more likely to display high levels of empathy. Oxytocin has also been associated with social affiliation, emotional responses, and love. "Rodrigues said previous research has shown that people with autism display lower scores on behavioral and dispositional empathy measures, and that a nasal spray with oxytocin increases scores in these areas." [Oregon] Though there is a very strong argument for the genetic basis of empathy due to its association with oxytocin, there are plenty of people with GG alleles who are also

empathetic. Once again, research proves a disposition towards a certain phenotype, not a guarantee.

Conclusion

The person we are today and the person we will become is not predestined in the “book of DNA.” We are predisposed to certain diseases, and we unfortunately sometimes inherit mutations that can lead to Huntington’s or other ultimate fates. Our phenotype, physical and behavioral, can be linked to what we carry in our four letter alphabet sequence and to what we experience as we try to survive. Sometimes only a small trigger is required to turn a gene on or off. Other times, a lifetime of choices can degrade our telomeres and DNA or deactivate a tumor suppressor gene. We cannot blame nurture or nature but simply accept that they work together. Understanding the conditions and contributions towards diseases, behaviors, and characteristics can ultimately personalize human medicine. Companies such as 23andMe and Navigenics have already begun this process by searching for predispositions for certain traits and diseases in customer’s DNA. They have developed a foundation for custom medical advice and treatments. However, environmental factors always come in to play. The future of medicine lays in the nature AND nurture argument – successful treatments will bear in mind a patient’s personal and genetic history.

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